Klinefelter Syndrome with Portal Vein Aneurysm: Case Report

Portal Ven Anevrizması Saptanan Klinefelter Sendromu Olgusu

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ABSTRACT

Klinefelter syndrome (KS) is the most common chromosomal disorder in men characterized by clinical features of hypogonadism and infertility. About 90% of cases have classically 47,XXY karyotype and the remaining have additional X or Y chromosomes, high grade aneuploidies or X chromosome structural abnormalities. Portal vein aneurysms are very rare. Reported cases are increasing due to use of modern imaging techniques in clinic practise. Here we report a 19-year-old man with KS who was admitted with complaints of abdominal pain, nausea and vomiting. Further investigations revealed 23 mm anechoic, saccular expansion in the left branch of the portal vein. It is well known that KS is associated with venous thromboembolic diseases including portal venous thrombosis, but association with portal vein aneurysm has not been previously reported.

Key Words: Portal vein aneurysm, klinefelter syndrome

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ÖZET

Klinefelter sendromu (KS), hipogonadizm ve infertilite ile karakterize, erkeklerde en yaygın görülen kromozomal bozukluktur. Olguların yaklaşık % 90'ında klasik 47,XXY karyotipi, geri kalanlarında ise ek X veya Y kromozomu, yüksek dereceli anöploidler veya X kromozomu yapısal anomalileri bulunmaktadır. Portal ven anevrizmaları çok nadirdir. Klinik pratikte modern görüntüleme tekniklerinin kullanımı ile birlikte, bildirilen vaka sayıları artmaktadır. Burada karın ağrısı, bulantı ve kusma şikayetleri ile başvuran 19 yaşındaki KS'li bir erkek hasta sunmaktayız. Yapılan incelemelerde; portal venin sol dalında 23 mm'lik, anekoik, sakküler genişleme saptanmıştır. KS'nin portal ven trombozu da dahil olmak üzere venöz tromboembolik hastalıklarla ilişkili olduğu iyi bilinmektedir; ancak portal ven anevrizması ile ilişkisi daha önce bildirilmemiştir.

Anahtar Sözcükler: Portal ven anevrizması, klinefelter sendromu

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INTRODUCTION

Klinefelter syndrome (KS) is the most common chromosomal disorder with an incidence of 1:500 male live births. It is characterized by clinical features of hypogonadism and infertility. About 90% of cases have classically 47,XXY karyotype and the remaining have additional X or Y chromosomes, high grade aneuploidies or X chromosome structural abnormalities (1).

Venous aneurysms (VAs) have been reported to occur in most major veins frequently in popliteal, jugular, or saphenous veins. Visceral VAs have been increasingly described in recent years probably due to increase in imagings. Portal vein aneurysm (PVA) is the most common site of visceral VA, representing fewer than 3% of all VAs (2-4). Less than 200 cases of PVAs have been reported since first discovered in 1956 by Barzilai and Kleckner (5,6). PVA is usually detected incidentally on imaging studies. Patients are frequently asymptomatic or have unrelated gastrointestinal complaints of abdominal pain or nausea that prompt abdominal imaging. Doppler ultrasound is the most widely used method for determining the size of an aneurysm and monitoring for expansion. Maximum portal vein diameter of 20 mm is the diagnostic standard for extrahepatic portal vein aneurysms and 9 mm is the diagnostic standard for intrahepatic ones (7).

In this report we describe a 19 year-old man with KS and incidentally detected PVA.

CASE REPORT

A 19-year-old man was admitted with the complaints of abdominal pain, nausea, and vomiting. He had recently been diagnosed with KS by chromosomal analysis, while being investigated for eunucoid appearance and gynecomastia. He had not yet received testosterone treatment. On physical examination, no abdominal tenderness was found, and liver and spleen was nonpalpable. Normal bowel sounds were present. The laboratory studies revealed: white blood cell counts, 6.600/mm^3; hemoglobin level, 14.6 g/dL; AST, 18 U/L (0-35), ALT, 9 U/L (0-45); GGT 20 U/L (2-22); ALP 111 U/L (30-120); amylase 40 U/L (28-100); total bilirubin 1,47 mg/dL (0,3-1,2); direct bilirubin 0,39 mg/dL (0-0,2); glucose 97 mg/dL (74-106); creatinine 0,64 mg/dL (0,31-0,7); albumine 4,72 g/dL (3,5-5,2); CRP 1 mg/L(0,01-5); FSH 52,62 mIU/mL(1,5-12,4); LH 23,11 mIU/mL (1,7-8,6); total testosterone 3,28 ng/ml(1,75-7,81); estradiol 21,59 pg/ml (7,6-43); TSH 0,69 µU/mL(0,51-4,3). Chest and abdominal radiographs were normal. An abdominal ultrasound scan showed 23 mm anechoic, saccular expansion in the left branch of the portal vein (Figure. 1). Monophasic, turbulent venous flow was detected in aneurysmal dilatation by doppler ultrasonography (Figure. 2). There were no features of thrombosis, portal hypertension, chronic liver disease, pancreatic mass or pancreatitis. No aneurysmal change was noted in the arterial tree and there were no pathological findings in other intraabdominal organs.

This case was presented as a poster presentation in 17th European Congress of Endocrinology in Ireland, Dublin; 2015.

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He was referred to perform oesophagogastroduodenoscopy and revealed pangastritis. His abdominal pain was attributed to gastritis.

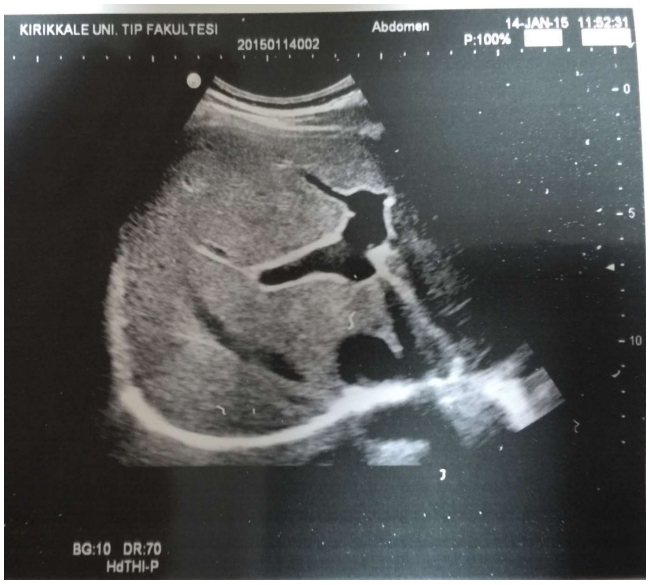

Figure 1. Anechoic, saccular expansion in the left branch of the portal vein

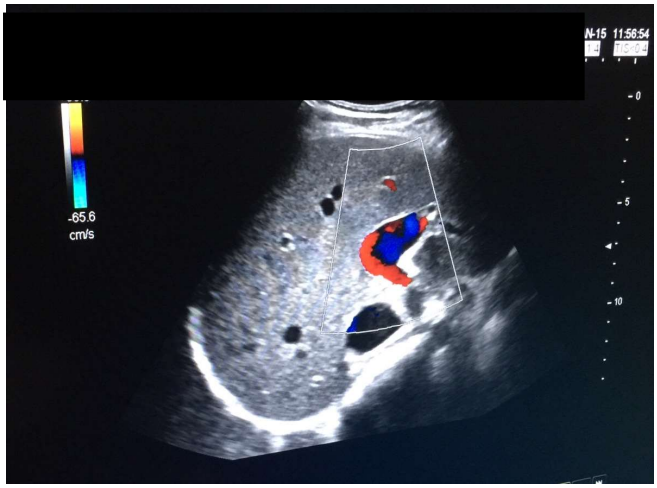

Figure 2. Doppler ultrasonography; monophasic, turbulent venous flow in aneurysmal dilatation

DISCUSSION

We have reported coexistance of KS and PVA in this paper. Patients with KS are at risk for a number of certain health problems including autoimmune disorders, venous thromboembolic disease, psychiatric disorders and connective tissue diseases (8,9). Intracranial aneurysms have been reported in KS (10,11). However; an association between KS and PVA was not previously reported. García González JP et al., previously reported a case of KS with atrial septal aneurysm. In this report; the aneurysm was thought to be related with connective tissue abnormality (12). Kasten R et al. and Ishihara K et al. previously reported cases of KS with mixed connective tissue disease and stated that low androgen level is the cause of connective tissue abnormality in KS (9,13).

PVAs are rare vascular anomalies. They are divided into two groups: acquired and congenital. Chronic liver disease, portal hypertension, pancreatitis, trauma and malignancies are included in acquired etiologies (14). Congenital PVA results from inherent weakness in the vessel wall. In our case, none of these acquired causes of PVA were present. The association of PVA and KS may be coincidental or related with connective tissue abnormalities. However the literature datas are not strong enough to declaire this.

CONCLUSION

The association of KS with aneurysms is not clearly determined however connective tissue abnormalities secondary to androgen defficiency are accused. PVAs are usually asymptomatic. However in KS; it is important to early diagnosis of PVA to follow up the patient for the risk of portal vein thrombosis and the other complications including aneurysmal rupture, compression to adjacent organs and portal systemic shunts. More detailed studies are needed to determine the relationship between PVA and CS.

Conflict of interest

No conflict of interest was declared by the authors.

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